



TNFRSF13B gene

TNF receptor superfamily member 13B

Normal Function

The *TNFRSF13B* gene provides instructions for making a protein called TACI. The TACI protein is found on the surface of immune system cells called B cells. These specialized white blood cells help protect the body against infection from foreign invaders such as bacteria and viruses. When B cells mature, they produce special proteins called antibodies (also known as immunoglobulins). Antibodies attach to specific foreign invaders, marking them for destruction. Through interactions with other proteins, TACI promotes cell signaling, plays a role in B cell survival and maturation, and is involved in the production of antibodies.

Health Conditions Related to Genetic Changes

common variable immune deficiency

More than 25 mutations in the *TNFRSF13B* gene have been associated with common variable immune deficiency (CVID). This condition impairs the immune system, resulting in increased risk for recurrent infections; autoimmune disorders, which occur when the immune system malfunctions and attacks the body's tissues and organs; and certain cancers.

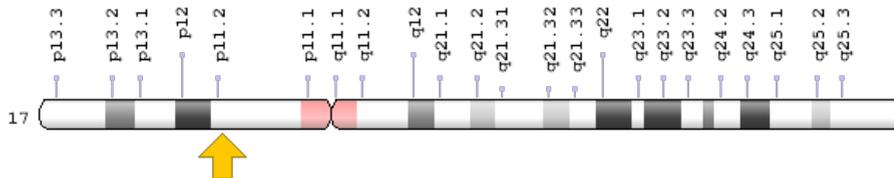
Most of the *TNFRSF13B* gene mutations associated with CVID change single protein building blocks (amino acids) in the TACI protein. The most common mutation seen in people with CVID replaces the amino acid cysteine with the amino acid arginine at position 104 in the TACI protein (written as Cys104Arg or C104R). This mutation impairs the ability of TACI to interact with other proteins, disrupting cell signaling and preventing normal B cell maturation and antibody production. A shortage (deficiency) of certain antibodies makes it difficult for people to fight off infections. Abnormal and deficient immune responses over time likely contribute to the increased cancer risk in people with CVID.

Some people with *TNFRSF13B* gene mutations do not develop the signs and symptoms of CVID. In these individuals, additional genetic or environmental factors are probably needed for the condition to occur.

Chromosomal Location

Cytogenetic Location: 17p11.2, which is the short (p) arm of chromosome 17 at position 11.2

Molecular Location: base pairs 16,939,084 to 16,972,088 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CD267
- IGAD2
- TACI
- TR13B_HUMAN
- transmembrane activator and CAML interactor
- tumor necrosis factor receptor 13B
- tumor necrosis factor receptor superfamily member 13B
- tumor necrosis factor receptor superfamily, member 13B

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Collection: TACI, BCMA
<https://www.ncbi.nlm.nih.gov/books/NBK6132/#A58801>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28TNFRSF13B%5BTIAB%5D%29+OR+%28TACI%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 13B
<http://omim.org/entry/604907>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TNFRSF13B.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TNFRSF13B%5Bgene%5D>
- HGNC Gene Family: CD molecules
<http://www.genenames.org/cgi-bin/genefamilies/set/471>
- HGNC Gene Family: Tumor necrosis factor receptor superfamily
<http://www.genenames.org/cgi-bin/genefamilies/set/782>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18153
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/23495>
- UniProt
<http://www.uniprot.org/uniprot/O14836>

Sources for This Summary

- Martinez-Gallo M, Radigan L, Almejún MB, Martínez-Pomar N, Matamoros N, Cunningham-Rundles C. TAC1 mutations and impaired B-cell function in subjects with CVID and healthy heterozygotes. *J Allergy Clin Immunol.* 2013 Feb;131(2):468-76. doi: 10.1016/j.jaci.2012.10.029. Epub 2012 Dec 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23237420>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3646641/>
- Pan-Hammarström Q, Salzer U, Du L, Björkander J, Cunningham-Rundles C, Nelson DL, Bacchelli C, Gaspar HB, Offer S, Behrens TW, Grimbacher B, Hammarström L. Reexamining the role of TAC1 coding variants in common variable immunodeficiency and selective IgA deficiency. *Nat Genet.* 2007 Apr;39(4):429-30.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17392797>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2931279/>
- Poodt AE, Driessen GJ, de Klein A, van Dongen JJ, van der Burg M, de Vries E. TAC1 mutations and disease susceptibility in patients with common variable immunodeficiency. *Clin Exp Immunol.* 2009 Apr;156(1):35-9. doi: 10.1111/j.1365-2249.2008.03863.x. Epub 2008 Dec 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19210517>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2673739/>

- Salzer U, Bacchelli C, Buckridge S, Pan-Hammarström Q, Jennings S, Lougaris V, Bergbreiter A, Hagen T, Birmelin J, Plebani A, Webster AD, Peter HH, Suez D, Chapel H, McLean-Tooke A, Spickett GP, Anover-Sombke S, Ochs HD, Urschel S, Belohradsky BH, Ugrinovic S, Kumararatne DS, Lawrence TC, Holm AM, Franco JL, Schulze I, Schneider P, Gertz EM, Schäffer AA, Hammarström L, Thrasher AJ, Gaspar HB, Grimbacher B. Relevance of biallelic versus monoallelic TNFRSF13B mutations in distinguishing disease-causing from risk-increasing TNFRSF13B variants in antibody deficiency syndromes. *Blood*. 2009 Feb 26;113(9):1967-76. doi: 10.1182/blood-2008-02-141937. Epub 2008 Nov 3.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18981294>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2651012/>
- Sathkumara HD, De Silva NR, Handunnetti S, De Silva AD. Genetics of common variable immunodeficiency: role of transmembrane activator and calcium modulator and cyclophilin ligand interactor. *Int J Immunogenet*. 2015 Aug;42(4):239-53. doi: 10.1111/iji.12217. Epub 2015 Jun 19. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/26096648>
- OMIM: TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 13B
<http://omim.org/entry/604907>
- Zhang L, Radigan L, Salzer U, Behrens TW, Grimbacher B, Diaz G, Bussel J, Cunningham-Rundles C. Transmembrane activator and calcium-modulating cyclophilin ligand interactor mutations in common variable immunodeficiency: clinical and immunologic outcomes in heterozygotes. *J Allergy Clin Immunol*. 2007 Nov;120(5):1178-85.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17983875>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2908504/>
- Zhang Y, Li J, Zhang YM, Zhang XM, Tao J. Effect of TACI signaling on humoral immunity and autoimmune diseases. *J Immunol Res*. 2015;2015:247426. doi: 10.1155/2015/247426. Epub 2015 Mar 17. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25866827>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4381970/>

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